Inequality of Sex Chromosome to Autosome Fusions

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Evolutionary Biology

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Abstract

Chromosomal fusions play an integral role in the remodeling of genomes and in karyotype evolution. Fusions that join a sex chromosome to an autosome are particularly abundant across the tree of life, but previous models on the establishment of such fusions have not accounted for the physical structure of the chromosomes. Our preliminary analysis predicts that a fusion joining an autosome to the pseudoautosomal region (PAR) of a sex chromosome will not remain stable, and the fusion will switch from the X to the Y chromosome each generation due to recombination. We have produced a forward time population genetic simulation to explore the outcomes of fusions to both the pseudoautosomal and non-recombining regions of sex chromosomes. The model can simulate the fusion of an autosome containing a sexually antagonistic locus to either the PAR or non-PAR end of a sex chromosome. This model is diploid, two-locus and biallelic, and is able to run thousands of simulations under a variety of conditions.

Sex Chromosome Systems

Fusions between autosomes and a sex chromosomes are one of the primary driving forces behind the establishment of Neo-XY and multiple sex chromosome systems. A substantial proportion of species of Mammalia, Coleoptera, and Polyneoptera have sex chromosome systems that resulted from autosome to sex chromosome fusions in the past.

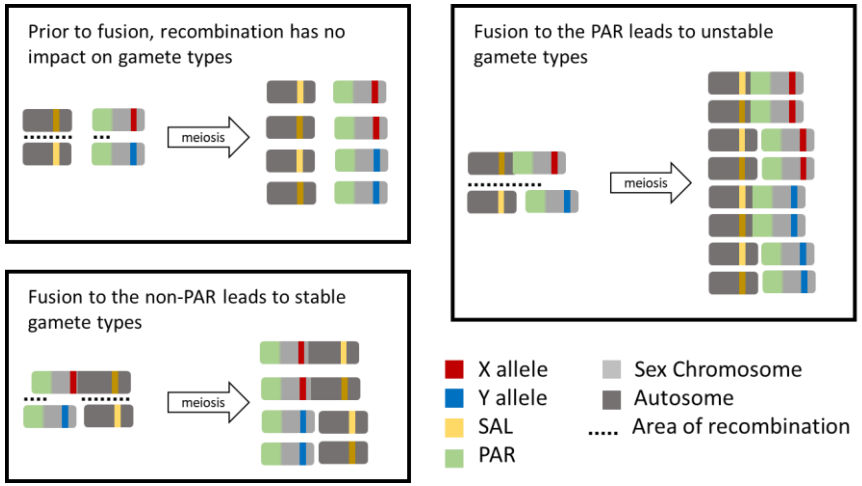
Despite the importance of autosome to sex chromosome fusions in reproductive isolation and sexual antagonism, the disproportionate number of autosome fusions to the Y chromosome cannot be adequately explained by a single model for the establishment of fusions. The current plausible explanations fail to take into account the potential effect that chromosomal structure and the size of the pseudoautosomal region (PAR) could have on the fate of sex chromosome to autosome fusions.

Clade	XO/XY	Fused Sex Chromosome System
Coleoptera	3870	411
Polyneoptera	512	311
Mammals	1626	202

Table 1: Ancestral and fused sex chromosome systems among clades

Fusions

The pseudoautosomal region, or PAR, is an area of the sex chromosome that can pair with the opposite sex chromosome and participate in recombination during meiosis. We explore the fate of four types of fusions: autosome to X PAR, autosome to X non-PAR, autosome to Y PAR, and autosome to Y non-PAR. It is important to note that when fusions occur with the PAR, male gametogenesis will often lead to production of a fused version of the alternative chromosome (Figure 1). We expect that a fusion to the non-PAR will result in stable inheritance of the fused sex chromosome system, while a fusion to the PAR will most likely switch between sex chromosomes every generation.



Model

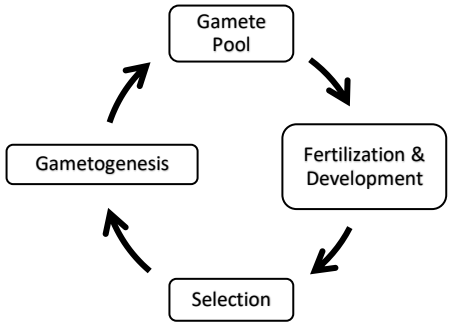
We developed a forward time population genetic simulation. Our model is diploid, two locus and biallelic. The first locus is on the sex chromosome and has alleles X and Y; individuals homozygous for the X allele are females while heterozygotes are male. This locus is in a non-recombining portion of the sex chromosomes, but each sex chromosome includes a recombining or pseduoautosomal region (PAR) as well. The second locus is a sexually antagonistic locus (SAL) and on an autosome and with alleles 0 and 1; 0 is beneficial to females while 1 is beneficial to males. The genetic architecture at this locus is described by the parameter h which represents dominance factor of the female benefit allele. The fitness of individuals is a function of their genotype at the SAL and their sex. We use a symmetrical model of sexual antagonism where the relative difference in fitness between sexes is equal (table 2).

Genotype	Male	Female
00	1+s	1
01	1+hs	1+(1-h)s
11	1	1+s

Table 2: Fitness function for males and females, where s is the selection coefficient and h is the dominance factor of the female beneficial allele (0).

Model

We first construct an initial population where alleles 0 and 1 are at equal frequency on autosomes and in males and females. Under the initial conditions no fusions are present in the population, and males and females are present in equal number. We then cycle through generations composed of four phases: selection, fusion mutations, gametogenesis, and fertilization. We implemented a model with viability selection within each sex where only 90% of individuals survive to mate but all survivors contribute equally to the gamete pool. During the fertilization step eggs are chosen at random and paired to randomly chosen X or Y bearing sperm to maintain a stable sex ratio and reconstitute the next generation of adults. Simulations are run until an equilibrium condition is reached (Figure 2).



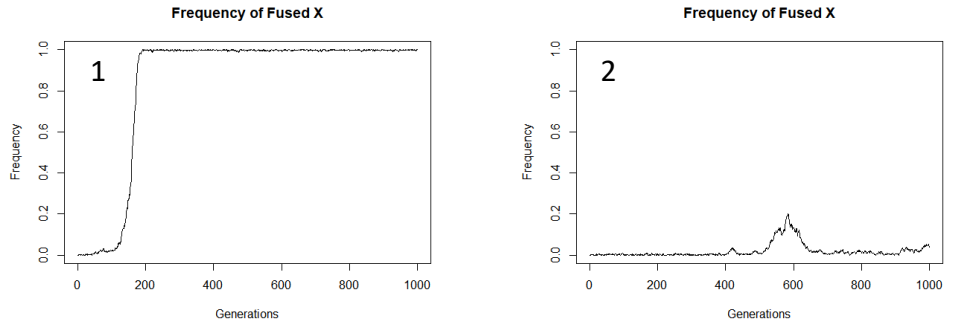


Figure 2 Possible fates of fusions: The frequency of a fusion of an autosome to the non-PAR of an X chromosome over 1000 generations in a population of 1000 individuals. After 1000 generations, (1) the fusion to the X chromosome has fixed in the population, and (2) the fusion has failed to fix in the population.

Recursion Equations

The stochastic model that has been described represents the fate of various chromosomal fusions in finite population over time. In order to further investigate the process, we have also created a series of recursion equations representing our model. This approach will reveal the fate of each type of fusion under an infinite population size model.

Expected outcomes

- Fusions to the non-PAR require a lower selection coefficient to fix than do fusions to the PAR.
- The closer to the fusion point that a SAL is located the more likely it is to drive a fusion to fixation.
- If the probability of a PAR or non-PAR fusions is significantly different among clades we should expect differences in the overall frequency of fusions that we observe.